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## **Echocardiographic study of cardiac rhabdomyomas from fetuses to children: about a tunisian hospital experience**

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# **Echocardiographic study of cardiac rhabdomyomas from fetuses to children: about a tunisian hospital experience**

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## **Abstract**

**Introduction.** Aim was to report our experience with regard to patients with cardiac rhabdomyomas , focusing on echocardiographic finding ,cardiac outcome and management.

**Material and methods.** Patients with cardiac rhabdomyomas diagnosis were retrospectively analyzed. during the period between 2000 and 2017.

**Results.** A total of 17 rhabdomyomas were diagnosed .seven patients were detected prenatally, six patients in the neonatal period and four patient in early infancy. , Tumors were mostly localized in the left ventricle (15/17) , their size ranged from 13\*10 mm to 55\*40 mm and it was obstructive in 7 cases. The tumors were singular in 8 patients and multiple in 9. We diagnosed tuberous sclerosis (TSC) in 9 babies. We reported 4 deaths , three patients required surgery because of hemodynamic obstruction in two cases and resistant arrhythmias in one baby. After a follow up of 45 months , 5 patients had marked tumor regression and 9 had complete tumor regression.

**Conclusions.** Cardiac rhabdomyoma may have different presentations and clinical course. It thought to be a benign tumor which tend to a spontaneous regression Surgery is only necessary when hemodynamically significant obstruction is present or resistant arrhythmia. As these tumors are associated with long-term development of TSC a follow-up was mandatory.

**Key words:** echocardiography, rhabdomyomas, fetuses

## **Introduction**

Cardiac rhabdomyoma is the most common pediatric cardiac tumor. Patients with cardiac rhabdomyoma are often asymptomatic or may present with murmur, arrhythmia, heart failure, or sudden death. Most cases are sporadic but a strong association with tuberous sclerosis complex (TSC) is reported in 30% to 50% of patients [1].

## **Objective**

The objective of this study is to report our experience on cardiac rhabdomyomas with cases diagnosed in our department and to put emphasis on echocardiographic findings , outcomes and on the disease's association with the signs of the tuberous sclerosis complex.

## Material and methods

We conducted a retrospective review of 17 patients aged 16 and under, who were diagnosed in the cardiopediatric department of the Rabta hospital with cardiac rhabdomyoma and who were admitted between 2000 and 2017.

Patients with benign cardiac tumors other than cardiac rhabdomyomas or malignant tumors or pseudo tumors such as hydatid cyst, thrombi, and endocarditis were excluded.

Clinical features of postnatal examinations were documented from records and echocardiographic images reviewed from the computer database. Data included age at diagnosis, clinical presentation, physical examination findings (cyanosis, heart murmur, arrhythmia, heart failure), electrocardiogram (ECG), 24-hour ECG recording results, initial and last echocardiography findings (number of rhabdomyomas, location, presence of inflow or outflow tract obstruction, myocardial dysfunction), indication for surgery, outcome (partial or total regression, residual tumors), and follow-up period.

The diagnosis of rhabdomyoma was based on echocardiographic aspects, eventual association with TSC, and on histologic examination in operated babies.

From 2000 to 2003 Echocardiographic imaging used Vingmed CFM 800 echocardiographic with poor resolution 5 to 8 MGz, then a VIIVD 5 and 9.

Babies were subjected to cardiac follow-up with clinical evaluation, color Doppler echocardiography and Holter-ECG every six months.

Complete tumor regression was defined as the complete disappearance of the tumor in echocardiography.

Partial tumor regression was defined as a decrease by at least 15% in tumor size.

Regarding patients with TSC, they were also under the supervision of a pediatric neurologist.

## Statistical analysis

Quantitative variables are expressed as means and standard deviations. Qualitative variables are expressed as percentages. Quantitative variables were expressed as means  $\pm$  standard deviations.

## Results

The baseline characteristics of patients was reported in **Table 1**. Seven cases of cardiac rhabdomyoma were diagnosed prenatally between 22 and 36 weeks of gestation with a mean of  $28 \pm 3.5$ . Six cases were diagnosed in the neonatal period and 4 cases in infancy.

Fetal tumors were monitored until birth. In three cases, tumors grew initially, reaching a mean size of 41x34 mm and caused an obstruction of the outflow tract. Then a mild regression was observed to the end of the 32nd week of gestation (Figures 1A and 1B). No complications were observed in utero.

In the other cases, tumors had a stable volume but changed in number (figure 2A and B). All babies were born full term and then transferred to the cardiology department, where the diagnosis was confirmed.

Postnatal diagnostic circumstances were diverse. Tumors were diagnosed incidentally in 3 cases, because of signs of heart failure ( $n = 7$ ), due to cardiac murmur ( $n = 4$ ), due to cyanosis, because of a tumor mimicking tricuspid atresia ( $n = 2$ ), and due to arrhythmia in only 1 case.

Eight patients had a single tumor while 9 had multiple tumors (Figure 3). Their sizes ranged from 13x10 mm to 55x40 mm. Masses were mostly localized in the left ventricle (in 15 out of the 17 cases) and were obstructive in 7 cases (Figure 4). Associated TSC was confirmed in 9 patients (53% of cases).

Four patients with cardiac rhabdomyoma in neonates died before surgery due to acute heart failure caused by obstruction.

All the remaining cases had been followed-up. A total tumor resection was performed in 4 patients because of cardiac failure due to an intracardiac obstruction in 3 cases and life-threatening resistant arrhythmias in one other case (Table 2).

Epileptic and neurological complications were observed in 9 patients.

After a median follow-up of 54 months, a complete or partial cardiac rhabdomyoma regression was observed in respectively 5 and 3 cases (Figure 5), and in one case it remained stable.

Patients with TSC had mental deficiency of variable degrees. Patients with seizures had received anticonvulsant medication under the care of pediatric neurologists.

## **Discussion**

Rhabdomyoma is the most common primary cardiac tumor in fetus and infants. It accounts for more than 60% of primary cardiac tumors [1].

Cardiac Rhabdomyoma is becoming more frequently diagnosed because of advances of ultrasonography as a part of the routine prenatal screening [2]. A neonatal diagnosis was performed during the second or third trimester [3, 4]. Similarly in our study, prenatal cases were detected at a mean of 28 weeks of gestation. Serial assessment of tumor size and its

haemodynamic effect throughout pregnancy is important in fact cardiac Rhabdomyomas increase in size until 32 weeks of gestation and then regress and become clinically less important [3]. Similar results were observed in our series since this progression and regression in size were detected in 3 fetus.

Cardiac rhabdomyoma is commonly associated with Tuberous sclerosis complex [5]. All neonates suspected of having rhabdomyoma need to be screened for (TS) as the association is between 50% and 80% [6, 7]. These numbers fit with our results, since TS correlates with cardiac rhabdomyomas in 53% of cases. rhabdomyomas associated to TSC are multiple in the majority of cases and can be located in any part of the heart. Ventricles are involved in most cases [8]. We got similar findings in our study.

The main treatment for cardiac rhabdomyoma is conservative management after infancy. In fact approximately ,more than 50% of the rhabdomyoma tumors showed a decrease in mass [9, 10]. We reached this result in our study when we observed partial and total spontaneous regression in 5 and 3 cases, respectively. There is no specific guidelines on how to follow up this group of patients [11]. Surgical tumor removal is considered in significant obstructive lesions with altered hemodynamic state or severe arrhythmia [12–14]. Similar results were reported in our study since surgery was indicated because of obstruction in 3 cases and resistant arrhythmias in one.

Cardiac rhabdomyomas may have a good short term prognosis. However, earlier Tuberous Sclerosis complex (TSC) may affect several organ systems, hindering neurodevelopment [14] and causing negative long term health outcomes.

## **Limitations**

The main limitations of our study are its retrospective nature and its small population size, both of which are due to the scarcity of infant cardiac tumor cases. Therefore, a multi-centric study is needed to corroborate our results.

Another limitation is the absence of a comparative study between cardiac rhabdomyomas, with or without TSc . Such a study would demonstrate the impact of this association on the prognosis of this type of tumor.

Despite this limitations , our study added a grow pool of data supporting the fact that rhabdomyomas has a variable clinical presentation and highlighted the role of prenatal echocardiographic detection and the trend of spontaneous regression of these types of tumors .

## **Conclusions**

Echocardiography is an essential tool for early diagnosis and follow-up of cardiac rhabdomyomas. These types of tumors trends to Have a positive outcomes which often tend to regress. All neonates with suspected rhabdomyomas need to be screened for (TSc) as this association determines the tumor's prognosis.

### Conflicts of interest

No conflicts of interest to be declared.

### References

1. Uzun O, Wilson DG, Vujanic GM, et al. Cardiac tumours in children. *Orphanet J Rare Dis.* 2007; 2: 11, doi: [10.1186/1750-1172-2-11](https://doi.org/10.1186/1750-1172-2-11), indexed in Pubmed: [17331235](https://pubmed.ncbi.nlm.nih.gov/17331235/).
2. Isaacs H. Fetal and neonatal cardiac tumors. *Pediatr Cardiol.* 2004; 25(3): 252–273, doi: [10.1007/s00246-003-0590-4](https://doi.org/10.1007/s00246-003-0590-4), indexed in Pubmed: [15360117](https://pubmed.ncbi.nlm.nih.gov/15360117/).
3. Fesslova V, Villa L, Rizzuti T, et al. Natural history and long-term outcome of cardiac rhabdomyomas detected prenatally. *Prenat Diagn.* 2004; 24(4): 241–248, doi: [10.1002/pd.825](https://doi.org/10.1002/pd.825), indexed in Pubmed: [15065096](https://pubmed.ncbi.nlm.nih.gov/15065096/).
4. Yuan SM. Fetal Primary Cardiac Tumors During Perinatal Period. *Pediatr Neonatol.* 2017; 58(3): 205–210, doi: [10.1016/j.pedneo.2016.07.004](https://doi.org/10.1016/j.pedneo.2016.07.004), indexed in Pubmed: [28043830](https://pubmed.ncbi.nlm.nih.gov/28043830/).
5. Sciacca P, Giacchi V, Mattia C, et al. Rhabdomyomas and tuberous sclerosis complex: our experience in 33 cases. *BMC Cardiovasc Disord.* 2014; 14: 66, doi: [10.1186/1471-2261-14-66](https://doi.org/10.1186/1471-2261-14-66).
6. Tworetzky W, McElhinney D, Margossian R, et al. Association between cardiac tumors and tuberous sclerosis in the fetus and neonate. *Am J Cardiol.* 2003; 92(4): 487–489, doi: [10.1016/s0002-9149\(03\)00677-5](https://doi.org/10.1016/s0002-9149(03)00677-5).
7. Józwiak S, Domańska-Pakieła D, Kwiatkowski D, et al. Multiple Cardiac Rhabdomyomas as a Sole Symptom of Tuberous Sclerosis Complex. *J Child Neurol.* 2016; 20(12): 988–989, doi: [10.1177/08830738050200121101](https://doi.org/10.1177/08830738050200121101).
8. Padalino MA, Vida VL, Bhattarai A, et al. Giant intramural left ventricular rhabdomyoma in a newborn. *Circulation.* 2011; 124(20): 2275–2277, doi: [10.1161/CIRCULATIONAHA.111.017897](https://doi.org/10.1161/CIRCULATIONAHA.111.017897), indexed in Pubmed: [22083149](https://pubmed.ncbi.nlm.nih.gov/22083149/).
9. Kohut J, Krzystolik-Ładzińska J, Szydłowski L, et al. The diagnosis, clinical course and follow-up of children with cardiac tumours - a single-centre experience. *Kardiol Pol.* 2010; 68(3): 304–309, indexed in Pubmed: [20411454](https://pubmed.ncbi.nlm.nih.gov/20411454/).

10. Chinawa J, Garekar S, Trivedi B. Large cardiac rhabdomyoma in two infants: Good response to medical management – A brief report. Nig J Cardiol. 2017; 14(2): 106–108, doi: [10.4103/njc.njc\\_19\\_17](https://doi.org/10.4103/njc.njc_19_17).
11. Hinton RB, Prakash A, Romp RL, et al. International Tuberous Sclerosis Consensus Group. Cardiovascular manifestations of tuberous sclerosis complex and summary of the revised diagnostic criteria and surveillance and management recommendations from the International Tuberous Sclerosis Consensus Group. J Am Heart Assoc. 2014; 3(6): e001493, doi: [10.1161/JAHA.114.001493](https://doi.org/10.1161/JAHA.114.001493), indexed in Pubmed: [25424575](https://pubmed.ncbi.nlm.nih.gov/25424575/).
12. Etuwewe B, John Cm, Abdelaziz M. Asymptomatic cardiac rhabdomyoma in neonates: is surgery indicated? Images Paediatr Cardiol. 2009; 11(2): 1–8, indexed in Pubmed: [22368553](https://pubmed.ncbi.nlm.nih.gov/22368553/).
13. Bejiqi R, Retkoceri R, Bejiqi H. Prenatally Diagnosis and Outcome of Fetuses with Cardiac Rhabdomyoma - Single Centre Experience. Open Access Maced J Med Sci. 2017; 5(2): 193–196, doi: [10.3889/oamjms.2017.040](https://doi.org/10.3889/oamjms.2017.040), indexed in Pubmed: [28507627](https://pubmed.ncbi.nlm.nih.gov/28507627/).
14. Chao AS, Chao A, Wang TH, et al. Outcome of antenatally diagnosed cardiac rhabdomyoma: case series and a meta-analysis. Ultrasound Obstet Gynecol. 2008; 31(3): 289–295, doi: [10.1002/uog.5264](https://doi.org/10.1002/uog.5264), indexed in Pubmed: [18307215](https://pubmed.ncbi.nlm.nih.gov/18307215/).
15. Lee KA, Won HS, Shim JY, et al. Molecular genetic, cardiac and neurodevelopmental findings in cases of prenatally diagnosed rhabdomyoma associated with tuberous sclerosis complex. Ultrasound Obstet Gynecol. 2013; 41(3): 306–311, doi: [10.1002/uog.11227](https://doi.org/10.1002/uog.11227), indexed in Pubmed: [22791573](https://pubmed.ncbi.nlm.nih.gov/22791573/).

**Table 1.** Baseline characteristis of population

Patients (n =)
Antenatal 7
Incidentally discovery 3
Cardiac murmur 4
Heart failure signs 7
Cyanosis 2
Arrhythmias 1

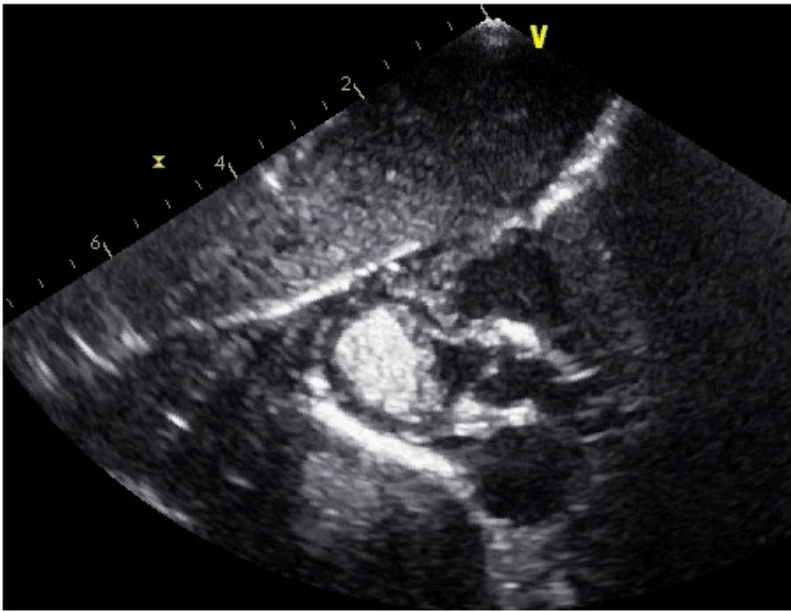
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**Table 2.** Characteristis of operatated patients

	<b>Age echocardiographic finding</b>	<b>Indication of surgery</b>	<b>Surgery and outcome</b>
<b>Patient1</b>	3 months multiples masses LV	Aortic outflow obstruction	Total excision + get well + no reccurence of tumors
<b>Patient2</b>	2 months giant single masse LV	Outflow obstruction	Total excision + get well + no reccurence of tumors
<b>Patients3</b>	20 days multiple masses LV+RV	Outflow obstruction	Total excision + get well + no reccurence of tumors
<b>Patient4</b>	4 months multiple masses LV	Severe and resistant arrythmias	Total excision + get well + no reccurence of arrythmias

LV — left ventricle; RV — right ventricle.





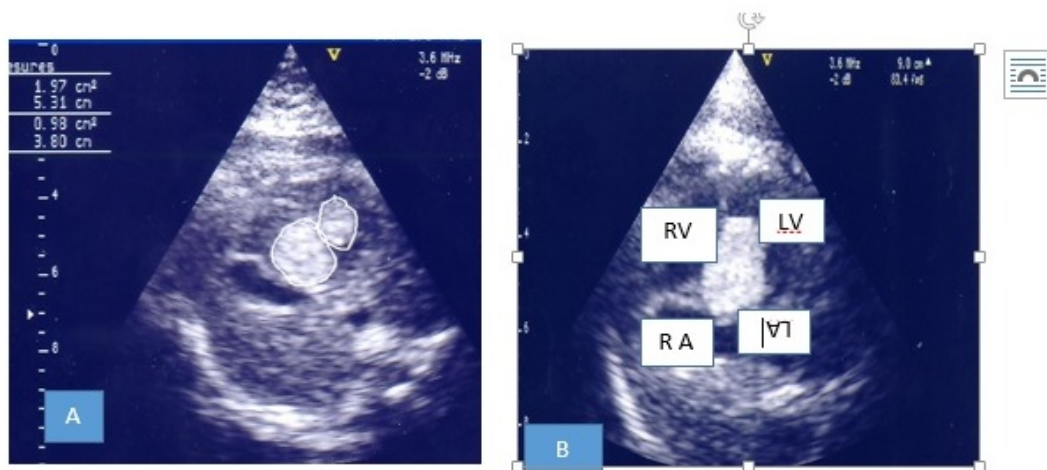


Figure 2A : fetal échocardiography (24weeks of gestations) : showed two masses localised in both left and right ventricle , 2B :a single mass tumor attached to ventricular septum and predominate in left ventricle



Figure 3 : echocardiography in a new born showed a a multiple rhabdomyomas

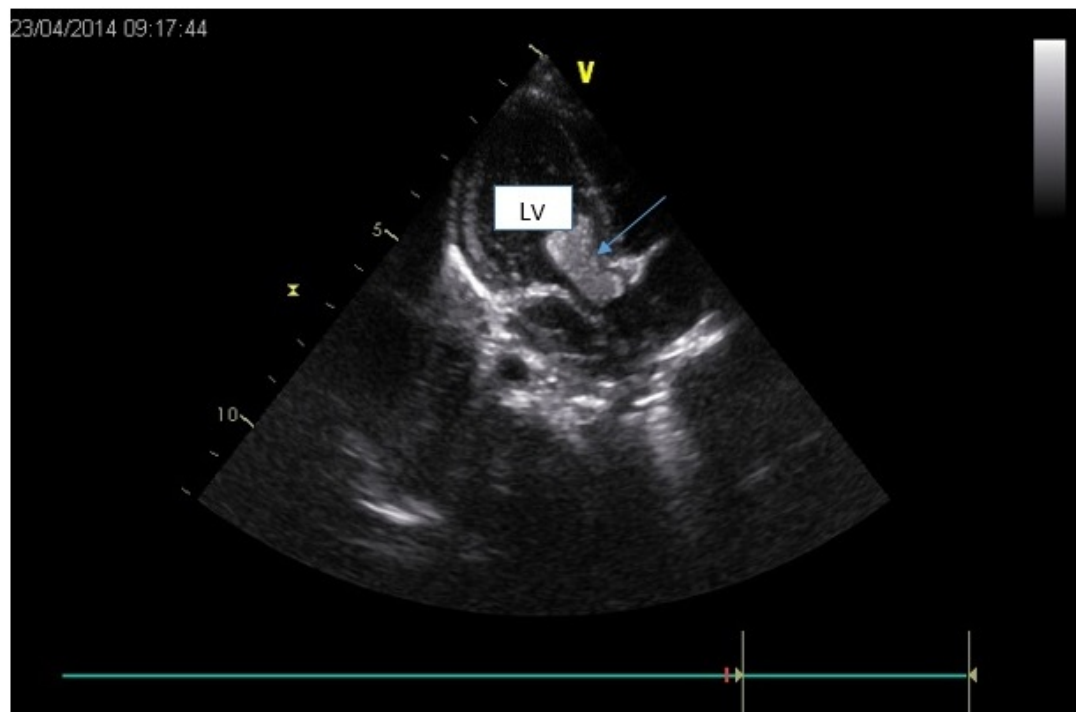


Figure4 : a multiple rhabdomyomas obstructing aortic outflow

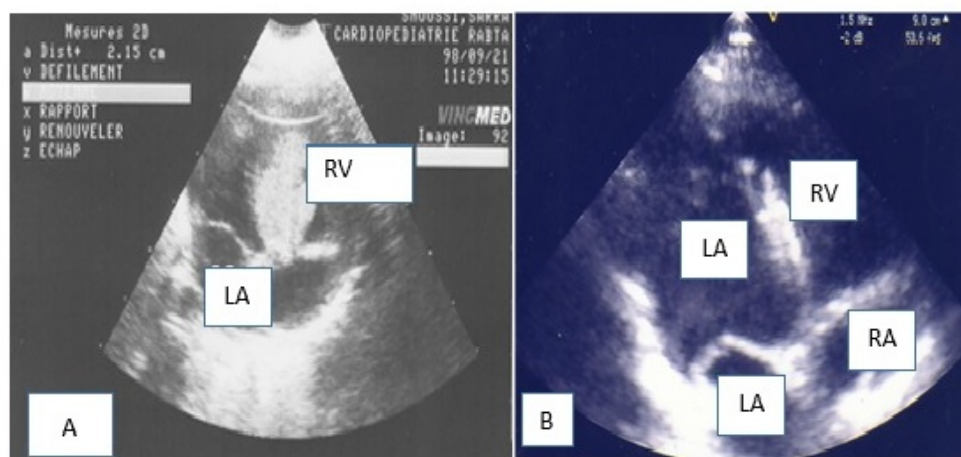


Figure 5A : right ventricular rhabdomyoma

Figure 5B : total regression of rhabdomyoma at a follow up of 45 months.